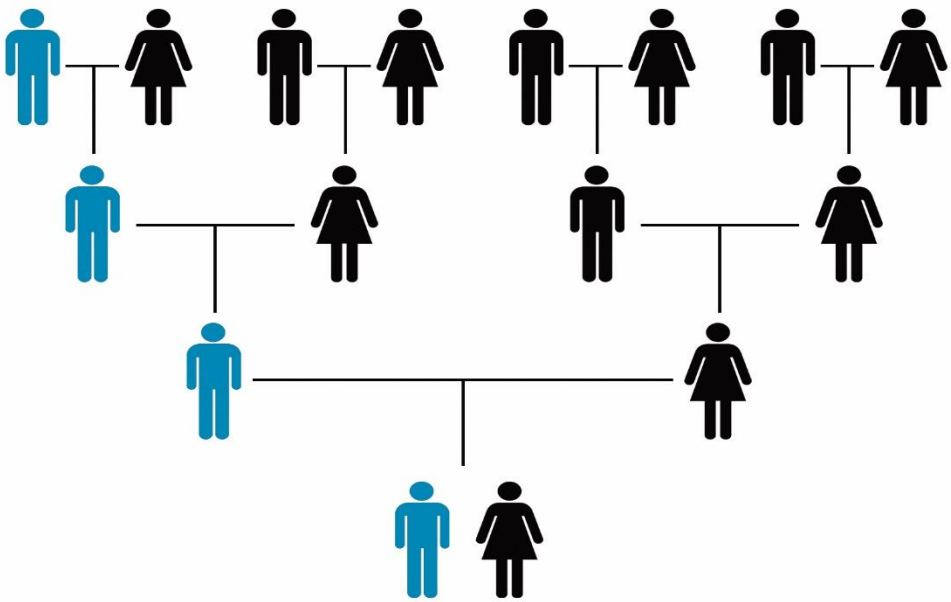
Chapter III. Genetics of diploid organisms



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Outlines of the Chapter III

- Mono-hybridism
- Di-hybridism : Independent segregation of two genes
- Poly-hybridism
- Gene linkage in diploid organisms
- Three-point test
- Epistasis
- Sex-linked inheritance

Objectives of this chapter

1. Understand mono-, di-, and poly-hybrid inheritance principles.

2. Analyze linkage and crossover events in diploid organisms.

3. Apply a three-point test for gene mapping.

4. Identify and classify epistatic interactions.

5. Explain and illustrate patterns of sex-linked inheritance.

1. Monohybridism

It is the study of the transmission of a single trait from one generation to another, through crossbreeding. A monohybrid cross is one in which both parents are **heterozygous** (or a **hybrid**) for a **single** (mono) trait.

A Punnett square and 3:1 ratio:

A Punnett square is a matrix in which all of the possible gametes produced by one parent are listed along one axis, and the gametes from the other parent are listed along the other axis.

Example: The F1 cross is shown in Figure 2. In a monohybrid cross, the offspring ratios will typically be 3:1, with three exhibiting the dominant phenotype (purple) and one exhibiting the recessive phenotype (white).

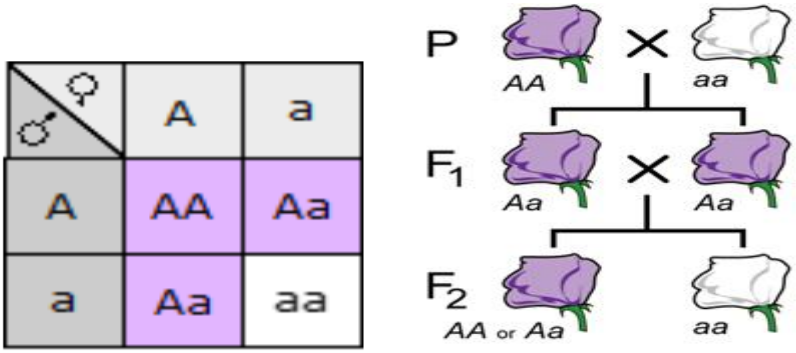
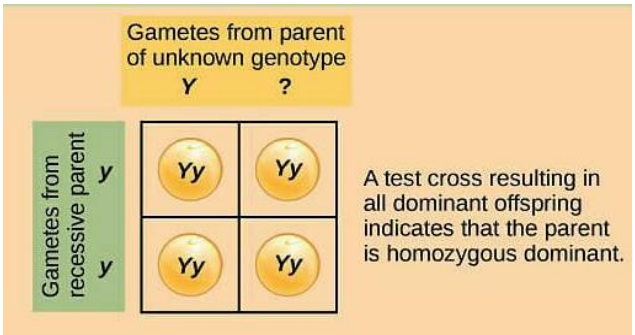
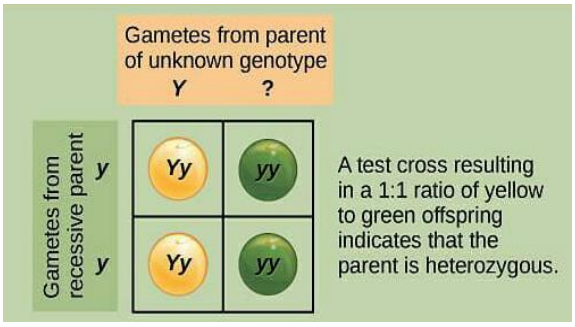


Fig. 2. A Punnett Square Showing a Monohybrid Cross.

- Because some alleles are dominant over others, the phenotype of an organism does not always reflect its genotype. A recessive phenotype (green) is only expressed with the organism is homozygous recessive (yy). A pea plant with yellow pods may be either homozygous (YY) or heterozygous (Yy). To determine whether an organism with a dominant phenotype (e.g. yellow pod color) is homozygous dominant or heterozygous, you use a **testcross**.

1.1.Test cross in monohybrid crosses

Test cross : it is the breeding of an organism of unknown genotype with a homozygous recessive. The results of this cross are shown in the table below.

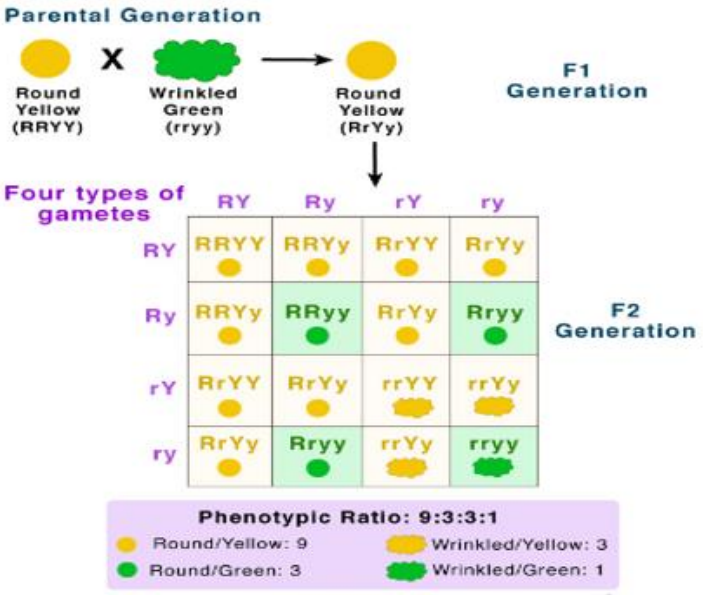
Possibility 1	**Possibility 2**
If all the progeny of the testcross have yellow pods, then the yellow pod parent was probably homozygous dominant since a YY x yy cross produces Yy progeny.	If the progeny of the testcross contains both green and yellow phenotypes, then the green pod parent was heterozygous since a Yy x yy cross produces Yy and yy progeny in a 1:1 ratio (50 percent for each of the two phenotypes).
	

Backcross: Backcrossing is a crossing of a hybrid with one of its parents or an individual genetically similar to its parent, in order to achieve offspring with a genetic identity which is closer to that of the parent.

2. Dihybridism

Refers to a cross between two distinct lines that differ in two observed traits. According to Mendel's law of independent assortment, genes for different traits can segregate independently during the formation of gametes.

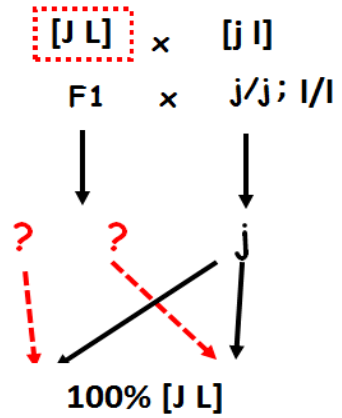
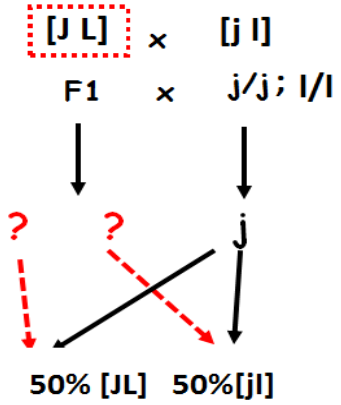
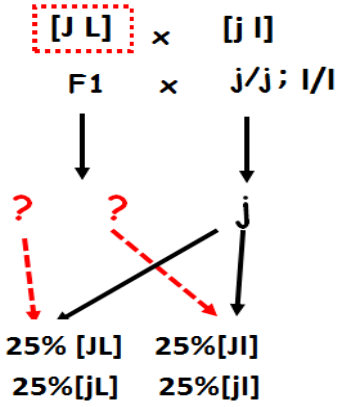
Example: considering the characters studied by Mendel "seed shape and seed color". The dihybrid crosses that Mendel performed consistently revealed the **9:3:3:1** ratio. Importantly, the F2 generation includes two phenotype classes (in this example, round, green peas and yellow, wrinkled peas) that are new combinations of phenotypes, distinct from the parental generation plants. Thus, independent assortment contribute to genetic diversity.



Using the **product rule**, we can multiply the individual probabilities of obtaining a round phenotype (¾) with the probability of obtaining a yellow phenotype (¾), then ¾ × ¾ = 9/16 of the progeny would be both round and green. Likewise, ¾ × ¼ = 3/16 of the progeny would be both round and yellow, and so on.

Product rule : The probability of two or more independent events occurring is the product obtained by multiplying the probabilities of the individual events. If you can use the word "and" to describe the occurrence of the events, then you can usually use the product rule. For example, the probability of an organism with phenotype 1 and phenotype 2 is the product of the probability of phenotype 1 times the probability of phenotype 2.

- ✓ **The test cross for studying the inheritance of two characteristics**: In order to distinguish genotypes from phenotypes expressing the dominant alleles, the test cross is essential. Three cases are possible: a homozygous individual dominant for the two traits, a heterozygous individual for one of the two traits or a heterozygous individual for both traits.

		
Dominant homozygote: In this case, the organism will possess two dominant alleles for a particular trait, and as a result, 100% of the offspring will express the dominant phenotype. This phenotype will consistently display the trait associated with the two dominant alleles.	**A simple heterozygote**: In the case of a heterozygote for one of the two traits, we expect to obtain two phenotypes with percentages of 50% each.	**A double heterozygote**: In the case of a heterozygote for both traits, we expect to obtain 4 phenotypes with percentages of 25%, 25%, 25%, and 25%.

3. Polyhybridism

Involves the study of inheritance patterns in crosses that consider three or more distinct traits. Each additional trait increases the complexity of the genetic combinations and the resulting phenotypic ratios.

Mendel's conclusions about the segregation of alleles and independent of assortment of genes continue to hold true for inheritance in diploid organisms.

The Product Rule can be used to predict outcomes when considering more than two genes at a time. Find the probability of each genotype or phenotype and multiple each probability.

Note : The number of phenotypes in a polyhybrid cross can be determined using the formula 2^n, where n is the number of traits being considered.

Example: Consider a cross involving three traits: seed shape, seed color, and flower position. If we denote the dominant and recessive alleles as follows:

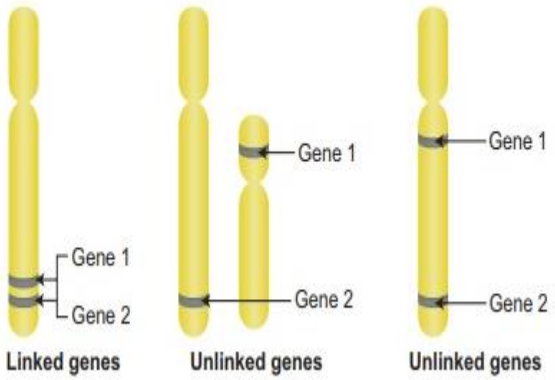
- Seed shape: Round (R) vs. Wrinkled (r)
- Seed color: Yellow (Y) vs. Green (y)
- Flower position: Axial (A) vs. Terminal (a)

The number of phenotypes is 2^3= 8. Using the product rule, the phenotypic ratio for a trihybrid cross (assuming independent assortment) is 27:9:9:9:3:3:3:1, where each ratio corresponds to a specific combination of the three traits.

Activity: If two organisms with genotype GgHhJi are crossed, what percent of offspring are expected to be homozygous dominant for all three genes? Hint: GG and HH and JJ

4. Linkage in diploid organisms

Refers to the localization of two or more genes on the same chromosome (linked genes). As a result, these genes remain together during the formation of gametes and are transmitted as a group. This behavior is in contrast to genes located on different chromosomes, which are transmitted independently. However, linked genes can still be separated and transmitted independently to varying degrees when a crossing-over event occurs.



Linkage can be either complete or incomplete:

- ✓ **Complete Linkage**: This occurs when linked genes do not separate. In this case, no recombinant phenotypes are observed; only the parental phenotypes appear.
- ✓ **Incomplete Linkage**: This occurs when genes can be separated by the phenomenon of crossing-over, but less frequently than independently assorting genes. In this scenario, both recombinant phenotypes will be observed in addition to the parental phenotypes, but at lower frequencies than if the genes were independent.

Example:

The linkage of genes is most often studied by the test cross; through its results, it tells us whether we're in the presence of independent or completely or incompletely linked genes. The examples bellow show the results obtained when in the case of complete linkage and incomplete linkage.

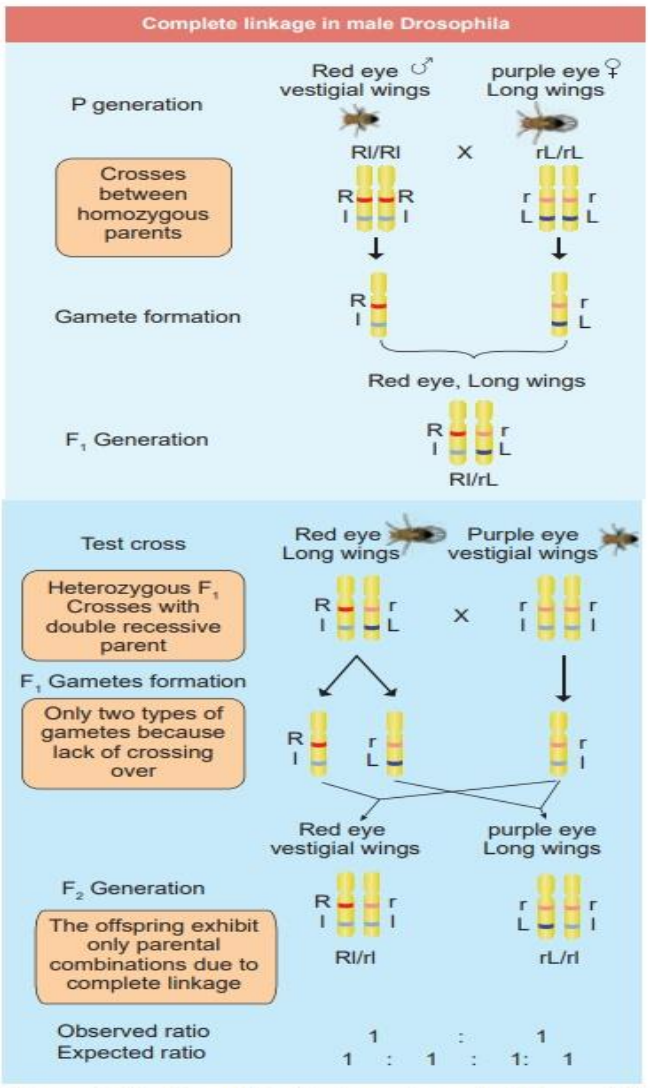


Figure 3.7: Complete linkage in male *Drosophila*

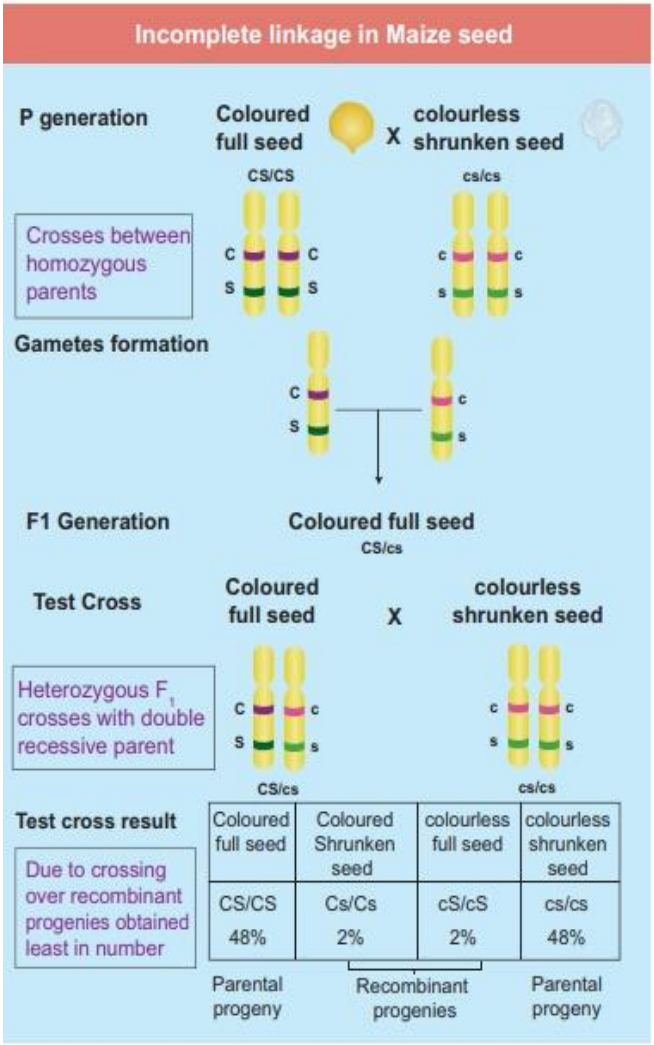


Figure 3.8: Incomplete linkage in Maize seed

Note: Genes that are incompletely linked can be distinguished from independent genes when the proportions of the four resulting phenotypes differ from a 1:1:1:1 ratio. Additionally, parental gametes appear more frequently than recombinant gametes (see figures below).

The distance between two genes on a chromosome in centimorgan = % of recombinant gametes produced following a crossover. In the example Cs/Cs+cS/cS=4%.

5. Three-point test

5.1.What is a three-point test cross?

In a three-point test cross, introducing a third gene allows for multiple types of crossover products to be observed. The figure below illustrates the various recombinant products that can result from these crossover events.

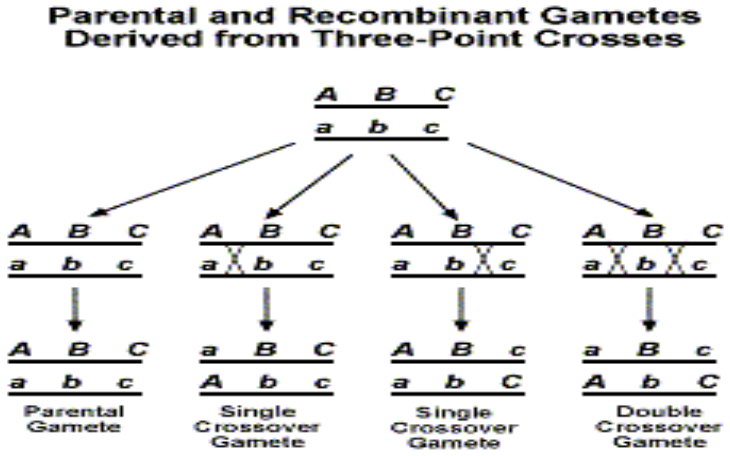


Fig.3. Recombinant products resulting from crossing-over in a three point cross.

Now if we were to perform a testcross with F1, we would expect a 1:1:1:1:1:1:1:1 ratio. As with the two-point analyzes described above, deviation from this expected ratio indicates that linkage is occurring.

5.2.Why do we need three-point test?

A three-point test cross allows geneticists to determine the order of three genes on a chromosome and the distances between them.

5.3.How to perform a three-point test?

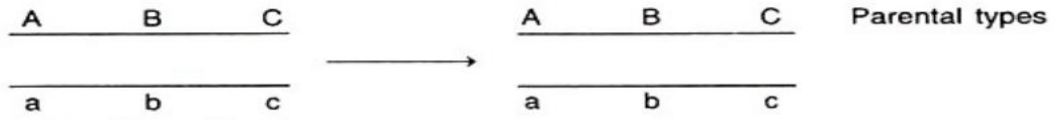
Let's take this example: the first step would be to cross a trihybrid heterozygous (F1) with a homozygous recessive parent, AaBbCc×aabbcc, then observe the offspring.

Example:

Genotype	Observed	Type of Gamete
ABC	390	Parental
abc	374	Parental
AbC	27	Single-crossover between genes *C* and *B*
aBc	30	Single-crossover between genes *C* and *B*
ABc	5	Double-crossover
abC	8	Double-crossover
Abc	81	Single-crossover between genes *A* and *C*
aBC	85	Single-crossover between genes *A* and *C*
Total	**1000**	

- ✓ **Step 1: determine parental genotypes**.

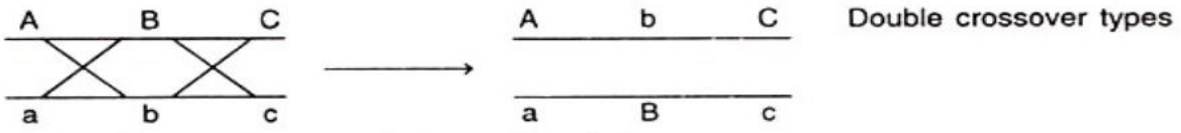
The genotypes found most frequently are the parental genotypes. From the table it is clear that the ABC and abc genotypes were the parental genotypes.



(*a*) No crossing over

- ✓ **Step 2: Determine the gene order**

Determine the double-crossover gametes (the genotypes with the lowest frequencies). In the example above the ABc and abC genotypes are the double crossovers. The double-crossover event moves the middle allele from one sister chromatid to the other. Thus, C gene must be in the middle because the recessive c allele is now on the same chromosome as the A and B alleles, and the dominant C allele is on the same chromosome as the recessive a and b alleles.

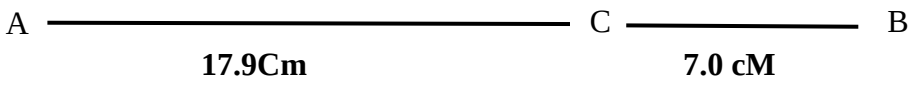


(*d*) Double crossing over between A and C

- ✓ **Step 3: Determining the linkage distances.**

The distance between genes A and C is calculated as follows: [100*((81+85+5+8)/1000)] = 17.9 cM, and the distance between C and B is [100*((27+30+5+8)/1000)] = 7.0 cM.

- ✓ **Step 4. Draw the map.**



6. Epistasis

6.1.Definition : Epistasis is a circumstance where the expression of one gene is modified (e.g., masked, inhibited or suppressed) by the expression of one or more other genes.

In epistasis the gene that does the masking is called **epistatic gene**, and the gene whose affect is masked is called **hypostatic gene**. Epistatic gene may be recessive or dominant in their effect.

6.2.Types of epistasis: types of gene interactions and their ratios are summarized in the table below.

Genotype	A- B- (AA BB, Aa BB, AABb, AaBb)	A- bb (AA bb, Aa bb)	Aa B- (aa BB, aa Bb)	aa bb
Classical ratio	9	3	3	1
Dominant epistasis	12		3	1
Recessive epistasis	9	3	4	
Duplicate Genes with Cumulative Effect	9	6		1
Duplicate Recessive Genes	15			1
Duplicate Dominant Genes	9	7		
Dominant and Recessive Interactions	13:3			

- ✓ **Dominant epistasis**: When a dominant allele at one locus can mask the expression of both alleles (dominant and recessive) at another locus, it is known as dominant epistasis. The obtained proportions will be (12:3:1).

Example:

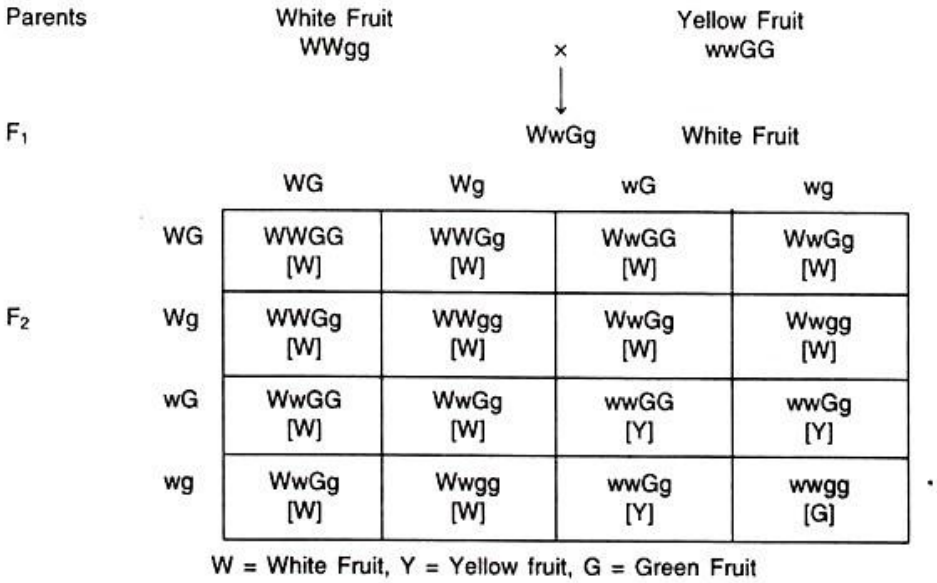
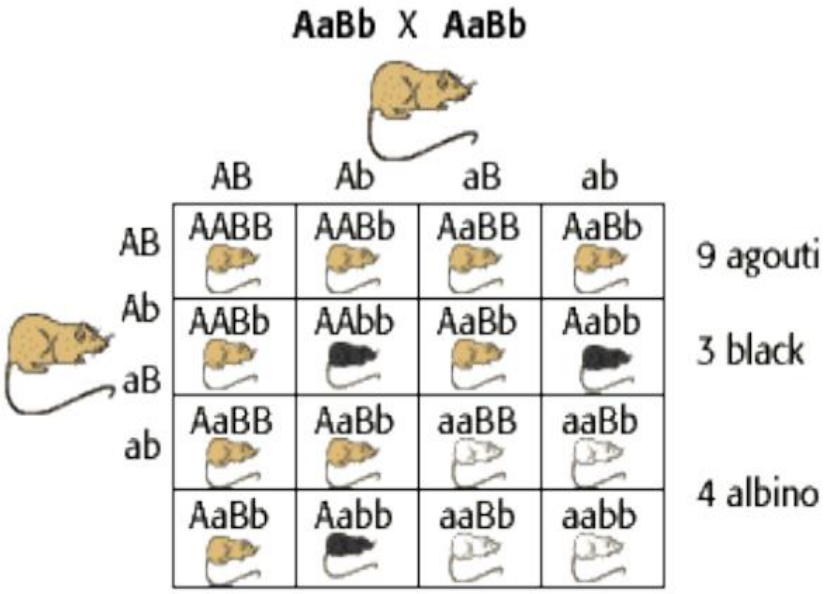


Fig. 8.3. Dominant epistasis for fruit colour in Summer squash. The normal dihybrid modified to 12 : 3 : 1 in F_2 generation.

- ✓ **Recessive epitasis or Supplementary gene action (9:3:4):** Sometimes the recessive alleles of one gene locus (aa) mask the action (phenotypic expression) of alleles of another gene locus (BB, Bb or bb alleles). This type of epistasis is called recessive epistasis.

Example:



7. **Sex-linked inheritance:** Transmission of hereditary traits linked to sex chromosomes (Gonosomes) XY.
   - ✓ **X-linked inheritance:** is performed by those genes which are localized in the non homologous sections of X chromosome, and that have no corresponding allele in Y chromosome. The X-linked genes are commonly known as sex-linked genes.
   - ✓ **Y-linked inheritance** : is performed by those genes which are localized in the non-homologous section of Y chromosome, and that have no alleles in X chromosome. (Holandric genes)
   - ✓ **XY-linked inheritance** : is performed by those genes which are localized in homologous sections of X and Y chromosomes

7.1. Characteristics of sex linked inheritance:

a) The pattern of inheritance of sex linked trait is **criss-cross**. It is the transmission of a gene from mother to son or father to daughter. Those patterns of inheritance are called **crisscross** inheritance or **skip generation** inheritance, in which a character is inherited to the second generation through the carrier of first generation.

b) The father cannot pass a sex linked allele to a son directly.

c) The mother can pass the allele of a trait to both daughter and son.

d) Only homozygous females can express a recessive trait, while heterozygous female are carriers and do not express the trait.

e) Males express the trait immediately because of the absence of a corresponding allele. This is the reason why males suffer from sex linked disorders more than females.

f) Most of the sex linked traits are recessive. Some examples of sex linked traits include Haemophilia or Bleeder's disease, Daltinism or Colour blindness.

7.2. Dominant inheritance: In this mode of transmission, the allele behaves as a dominant trait and occurs in both hemizygous (XY) males and heterozygous (XX) females (often to a lesser degree of severity).

- ✓ The affected males pass the condition on to all of their daughters but to none of their sons
- ✓ Females usually pass the condition (defective phenotype) on to one-half of their sons and daughters.
- ✓ X-linked dominant gene fails to be transmitted to any son from amother which did not exhibit the trait itself.

If the trait is related to X of the mother:

Parents	$X^A X^A \times X^a Y$
Gametes	X^A X^a , Y
Offspring	$X^A X^a$, $X^A Y$
100% of females [A] and 100% of males [A]	

Parents	$X^A X^a \times X^a Y$
Gametes	X^A , X^a X^a , Y
Offspring	$X^A X^a$, $X^a X^a$; $X^a Y$, $X^A Y$
50% of females [A] and 50% are [a] 50% of males [A] and 50% are [a]	

If the trait is related to X of the father :

Parents	$X^a X^a \times X^A Y$
Gametes	X^a X^A , Y
Offspring	$X^A X^a$, $X^a Y$
100% of females [A] and 100% of males [a]	

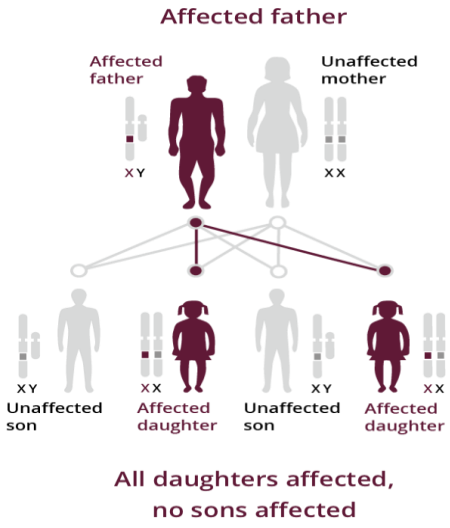
Example : X-Linked Hypophosphatemia (XLH)

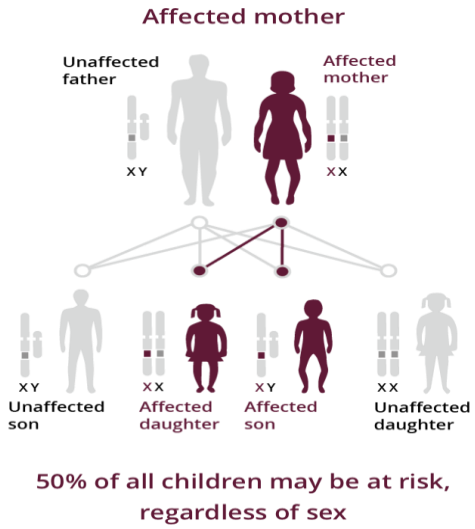
Cause: caused by mutations in the PHEX gene, located on the X chromosome. This mutation leads to impaired phosphate metabolism, causing low phosphate levels (hypophosphatemia) in the blood.

Characteristics:

- Weak, soft bones (rickets).
- Bowed legs.
- Short stature.
- Dental issues (abscesses).

Inheritance Pattern:





7.3.Recessive inheritance : X-linked recessive inheritance is a way a genetic trait or condition can be passed down from parent to child through mutations (changes) in a gene on the X chromosome. In this mode of inheritance, the allele behaves as a recessive trait. In females, it is expressed only in the homozygous state. However, the trait is always present in males (XY) who possess only one copy of the gene (hemizygotes).

- ✓ If only the father or the mother has the mutated X-linked gene, the daughters are usually not affected and are called carriers because one of their X chromosomes has the mutation but the other one is normal.
- ✓ Sons will be affected if they inherit the mutated X-linked gene from their mother. Fathers cannot pass X-linked recessive conditions to their sons.

Trait is related to X of the mother

Parents $X^AX^a \times X^AY$

Gametes X^A X^a X^A Y

Offspring X^AX^A , X^aX^A , X^AY, X^aY

100% of females [A]
50% of males [A] and 50% are [a]

Trait is related to X of the Father

Parents $X^AX^A \times X^aY$

Gametes X^A X^a Y

Offspring X^AX^a , X^AY

100% of females are [A]
100% of males are [A]

Example : Color Blindness

Cause : mutations in the OPN1LW or OPN1MW genes located on the X chromosome.

These genes are responsible for producing light-sensitive proteins in the retina that detect red and green light.

Inheritance pattern